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Proposed ASHG Position on Mapping/Sequencing the Human Genome

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Recent proposals to undertake a nationally focused effort to map/sequence the human genome have captured the political and media imaginations. Sound policy positions must be advanced to guide and focus such an effort. Upcoming decisions about organizational structure and allocation of resources may have profound effects on future research in human genetics. The ASHG Public Policy Committee determined that ASHG should make policy recommendations in key areas related to this project.

The usual process of formation of an ASHG expert committee to develop society policy recommendations was not undertaken since the National Academy of Sciences was known to be organizing a task force to develop scientific consensus on federal policy for a human genome project. Since this NAS committee contained many ASHG members and worked in the same time frame, the Public Policy Committee anticipated that its key recommendations could serve as a draft policy position for review by ASHG. Chaired by Bruce M. Alberts, the committee included members David Botstein, Sydney Brenner, Charles R. Cantor, Russell F. Doolittle, Leroy Hood, Victor McKusick, Daniel Nathans, Maynard V. Olson, Stuart Orkin, Leon Rosenberg, Francis H. Ruddle, Shirley Tilghman, John Tooze, and James D. Watson.

The key recommendations of the recently published report of this committee on mapping and sequencing the human genome are summarized below. ASHG members are asked to provide comment to the Public Policy Committee on the proposal that similarly worded statements become the policy positions of the ASHG. These recommendations address the five key areas in which responsible advice from the scientific community is needed: the appropriate scale, order, and progression of the scientific effort; recommended federal funding mechanisms; the scientifically appropriate balance of centralized and decentralized research; the new facilities and resources needed; and the recommended locus for leadership of the program.

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The Committee on Mapping and Sequencing the Human Genome of the Board on Basic Biology of the National Academy of Sciences made the following policy recommendations (Executive Summary):

- Acquiring a map, a sequence, and an increased understanding of the human genome merits a special effort that should be organized and funded specifically for this purpose. Such a special effort in the next two decades will greatly enhance progress in human biology and medicine.

- The technical problems associated with mapping and sequencing the human and other genomes are sufficiently great that a scientifically sound program should begin with a diversified, sustained effort to improve our ability to analyze complex DNA molecules. Although the needed capabilities do not yet exist, the broad outlines of how they could be developed are clear. Prospects are therefore good that the required advanced DNA technologies would emerge from a focused effort that emphasizes pilot projects and technological development. Once established, these technologies would not only make the complete analysis of the human and other genomes feasible, but would also revolutionize many other areas of basic biology and biotechnology.

- Important early goals of the effort should be to acquire a high-resolution genetic linkage map of

the human genome, a collection of ordered DNA clones, and a series of complementary physical maps of increasing resolution. The ultimate goal would be to obtain the complete nucleotide sequence of the human genome, starting from the materials in the ordered DNA clone collection. Attaining this goal would require major (but achievable) advances in DNA handling and sequencing technologies.

- A comparative genetic approach is essential for interpreting the information in the human genome. Therefore, intensive studies of those organisms that provide particularly useful models for understanding human gene structure and function and evolution must be carried out in parallel.

- The mapping and sequencing effort should begin primarily as a series of competing, peer-reviewed programs emphasizing technology development. Funding should include both grants to individuals and grants to medium-sized multidisciplinary groups of scientists and engineers. Because the technology required to meet most of the project's goals needs major improvement, the committee specifically recommends against establishing one or a few large sequencing centers at present.

- The human genome project should differ from present ongoing research inasmuch as the component subprojects should have the potential to improve by 5- to 10-fold increments the scale or efficiency of mapping, sequencing, analyzing, or interpreting the biological significance of the information in the human genome.

- Progress toward all the above goals will require the establishment of well-funded centralized facilities, including a stock center for the cloned DNA fragments generated in the mapping and sequencing effort and a data center for the computer-based collection and distribution of large amounts of DNA sequence information. The committee suggests that the groups supplying these services be selected through open competition.

On the basis of these conclusions, the committee recommends the following:

- In view of the importance and magnitude of the task, a rapid scale-up to $200 million of additional funding per year is recommended. These additional funds should not be diverted from the current federal research budget for biomedical sciences.

A majority of the committee recommends that

- A single federal agency serve as the lead agency for the project. This agency would receive and administer the funds for the project and would be responsible for the operation of the stock center and data center, as well as administer the peer review system utilized in determining the recipients of funds. It should work closely with a Scientific Advisory Board in developing and implementing a high standard of peer review. The Scientific Advisory Board, composed primarily of scientists knowledgeable in relevant fields, would provide advice not only on peer review, but also on quality control, international cooperation, coordination of efforts of the laboratories in the project, and the stock and data centers.

In addition, the ASHG Public Policy Committee would like comment on whether the ASHG position statement should include

- a recommendation that NIH be the lead agency for coordination of the national research effort;

- a recommendation that an effort of this magnitude must include research training resources to meet the need for appropriately trained scientists as the project progresses;

- an expression of belief that the human genome sequences should be a public trust and therefore not subject to copyright.

Comments should be addressed to the Public Policy Committee, American Society of Human Genetics, 9650 Rockville Pike, Bethesda, MD 20814 before September 10, 1988.